

TRANSMITTAL  
FORM

(to be used for all correspondence after initial filing)

Total Number of Pages in This Submission

7

Application Number	10/526,429
Filing Date	February 28, 2005
First Named Inventor	ROMMENS, Johanna M.
Art Unit	TBD
Examiner Name	TBD
Attorney Docket Number	8092-002-US

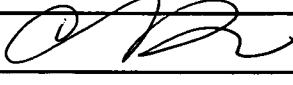
ENCLOSURES (Check all that apply)		
<input type="checkbox"/> Fee Transmittal Form	<input type="checkbox"/> Drawing(s)	<input type="checkbox"/> After Allowance Communication to TC
<input type="checkbox"/> Fee Attached	<input type="checkbox"/> Licensing-related Papers	<input type="checkbox"/> Appeal Communication to Board of Appeals and Interferences
<input type="checkbox"/> Amendment/Reply	<input type="checkbox"/> Petition	<input type="checkbox"/> Appeal Communication to TC (Appeal Notice, Brief, Reply Brief)
<input type="checkbox"/> After Final	<input type="checkbox"/> Petition to Convert to a Provisional Application	<input type="checkbox"/> Proprietary Information
<input type="checkbox"/> Affidavits/declaration(s)	<input type="checkbox"/> Power of Attorney, Revocation	<input type="checkbox"/> Status Letter
<input type="checkbox"/> Extension of Time Request	<input type="checkbox"/> Change of Correspondence Address	<input type="checkbox"/> Other Enclosure(s) (please Identify below):
<input type="checkbox"/> Express Abandonment Request	<input type="checkbox"/> Terminal Disclaimer	<input type="checkbox"/> Self Addressed Pre-Paid Stamped Postcard
<input checked="" type="checkbox"/> Information Disclosure Statement	<input type="checkbox"/> Request for Refund	
<input type="checkbox"/> Certified Copy of Priority Document(s)	<input type="checkbox"/> CD, Number of CD(s) _____	
<input type="checkbox"/> Reply to Missing Parts/ Incomplete Application	<input type="checkbox"/> Landscape Table on CD	
<input type="checkbox"/> <input type="checkbox"/> Reply to Missing Parts under 37 CFR 1.52 or 1.53		
Remarks		

## SIGNATURE OF APPLICANT, ATTORNEY, OR AGENT

Firm Name	CATALYST LAW GROUP, APC		
Signature			
Printed name	David M. Kohn, J.D.		
Date	February 9, 2006	Reg. No.	53,150

## CERTIFICATE OF TRANSMISSION/MAILING

I hereby certify that this correspondence is being facsimile transmitted to the USPTO or deposited with the United States Postal Service with sufficient postage as first class mail in an envelope addressed to: Commissioner for Patents, P.O. Box 1450, Alexandria, VA 22313-1450 on the date shown below:

Signature	
Typed or printed name	Christina Dueñas
Date	February 9, 2006

This collection of information is required by 37 CFR 1.5. The information is required to obtain or retain a benefit by the public which is to file (and by the USPTO to process) an application. Confidentiality is governed by 35 U.S.C. 122 and 37 CFR 1.11 and 1.14. This collection is estimated to 2 hours to complete, including gathering, preparing, and submitting the completed application form to the USPTO. Time will vary depending upon the individual case. Any comments on the amount of time you require to complete this form and/or suggestions for reducing this burden, should be sent to the Chief Information Officer, U.S. Patent and Trademark Office, U.S. Department of Commerce, P.O. Box 1450, Alexandria, VA 22313-1450. DO NOT SEND FEES OR COMPLETED FORMS TO THIS ADDRESS. SEND TO: Commissioner for Patents, P.O. Box 1450, Alexandria, VA 22313-1450.

If you need assistance in completing the form, call 1-800-PTO-9199 and select option 2.



February 9, 2006

CERTIFICATE OF MAILING  
(37 C.F.R. §1.8a)

I hereby certify that this paper (along with any referred to as being attached or enclosed) is being deposited with the United States Postal Service on the date shown below with sufficient postage as First Class Mail in an envelope addressed to the Commissioner for Patents, Mail Stop: Information Disclosure Statement, P. O. Box 1450, Alexandria, VA 22313-1450.

Christina Dueñas  
Name of Person Mailing Paper

Signature of Person Mailing Paper

February 9, 2006  
Date of Deposit

**IN THE UNITED STATES PATENT AND TRADEMARK OFFICE**

In re Application of:	)	Examiner: TBA
	)	
Rommens, Johanna M., et al.	)	Group Art Unit: TBA
	)	
Serial No.: 10/526,429	)	Docket No.: 8092-002-US
	)	
Filed: February 28, 2005	)	Date Mailed: February 9, 2006
	)	
For: DIAGNOSIS OF SHWACHMAN-	)	
DIAMOND SYNDROME	)	
	)	

**INFORMATION DISCLOSURE STATEMENT**

Mail Stop: Information Disclosure Statement  
Honorable Commissioner for Patents  
P.O. Box 1450  
Alexandria, VA 22313-1450

Dear Commissioner:

This document is an Information Disclosure Statement to the above-cited patent application and filed pursuant to 37 C.F.R. § 1.97(b)(3). Therefore, no fee is believed to be due.

Attached is form 1449A/PTO listing documents believed relevant to the subject application. The submission of this document is not intended, nor should it be construed, to constitute an admission that any patent, patent application, article, or other information referred to herein is "prior art" unless specifically designated as such. In fact, applicants submit that these documents do not affect the patentability of the subject invention. In accordance with 37 C.F.R. § 1.97(g), the filing of this information shall not be construed to represent that a search has been made or that no other material information may exist. Neither should its submission be construed to indicate that a thorough search should not be conducted by the Examiner.

It is respectfully requested that the listing of documents in the attached forms be: (1) fully considered by the Patent and Trademark Office during the prosecution of this application; and (2) represented on any patent which may issue on the application. Applicant respectfully requests that the copies of the forms 1449A/PTO, be considered and initialed by the Examiner, be returned with the next communication pursuant to the §609 of the Manual of Patent Examining Procedures (MPEP).

The order of the documents listed in the attached forms is to be accorded no particular import, as the order thereof is completely fortuitous. Additionally, the documents are not necessarily analogous art. A copy of each article document listed in the attached form is enclosed.

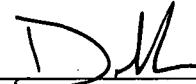
It is believed that this disclosure complies with the requirements as set forth under 37 C.F.R. § 1.56, § 1.97, and § 1.98 and §707.05(b) of the MPEP. If for some reason the Examiner considers otherwise, it is respectfully requested that the undersigned be telephoned at (858) 450-0099 so that any deficiencies can be remedied.

It is further believed that this Information Disclosure Statement is being submitted before the mailing of an Office Action on the merits. Accordingly, no fee is believed to be due

under 37 C.F.R. § 197(b)(3). If an Office Action on the merits has in fact been mailed, authorization is hereby given to charge the required fee of \$180.00 under 37 C.F.R. § 1.97(c)(2) and 1.17(p) to Deposit Account No. 502235.

Respectfully submitted,

Date: 2/9/04

  
\_\_\_\_\_  
David M. Kohn, J.D.  
Reg. No. 53,150

CATALYST LAW GROUP, APC  
9710 Scranton Road, Suite 170  
San Diego, California 92121  
(858) 450-0099  
(858) 450-9834 (Fax)

Substitute form 1449A/PTO		<i>Complete if Known</i>	
<b>INFORMATION DISCLOSURE STATEMENT BY APPLICANT</b> <i>(use as many sheets as necessary)</i>		Application Number	10/526,429
		Filing Date	February 28, 2005
		First Named Inventor	ROMMENS, Johanna M.
		Group Art Unit	TBD
		Examiner Name	TBD
		Attorney Docket Number	8092-002-US

U.S. PATENT APPLICATIONS				
Examiner Initials*	Cite No.	U.S. Serial No.	Name of Applicant of Cited Document	Date of Filing of Cited Document MM-DD-YYYY
		US-		

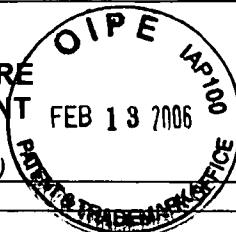
OTHER NON PATENT LITERATURE DOCUMENTS				
Examiner Initials*	Cite No.	Include name of the author (in CAPITAL LETTERS), title of the article (when appropriate), title of the item (book, magazine, journal, serial, symposium, catalog, etc.), date, page(s), volume-issue number(s), publisher, city and/or country where published		
/D.T./	2.	SHWACHMAN, H., Diamond, L.K. & Khaw, K., (1964), "The Syndrome of Pancreatic Insufficiency and Bone Marrow Dysfunction", J. Pediatr. v. 65, pp. 645-663.		
/D.T./	3.	GINZBERG, H. et al., (1999), "Shwachman Syndrome: Phenotypic Manifestations of Sibling Sets and Isolated Cases in a Large Patient Cohort are Similar", J. Pediatr. v. 135, pp. 81-88.		
Examiner Signature		/David Thomas/	Date Considered	05/27/2008

\*EXAMINER: Initial if reference considered, whether or not citation is in conformance with MPEP 609. Draw line through citation if not in conformance and not considered. Include copy of this form with next communication to applicant.

INFORMATION DISCLOSURE  
STATEMENT BY APPLICANT

(use as many sheets as necessary)

Sheet 2 of 3



Complete if Known	
Application Number	10/526,429
Filing Date	February 28, 2005
First Named Inventor	ROMMENS, Johanna M.
Group Art Unit	TBD
Examiner Name	TBD
Attorney Docket Number	8092-002-US

## OTHER NON PATENT LITERATURE DOCUMENTS

Examiner Initials*	Cite No.	Include name of the author (in CAPITAL LETTERS), title of the article (when appropriate), title of the item (book, magazine, journal, serial, symposium, catalog, etc.), date, page(s), volume-issue number(s), publisher, city and/or country where published	T
/D.T./	4.	GINZBERG, H. et al., (2000), "Segregation analysis in Shwachman-Diamond Syndrome: Evidence for Recessive Inheritance", Am. J. Hum. Genet, v. 66, pp. 1413-1416.	
	5.	GOOBIE, S. et al., (2001), "Shwachman-Diamond syndrome with exocrine pancreatic dysfunction and bone marrow failure maps to the centromeric region of chromosome 7", Am. J. Hum. Genet, v. 68, pp. 1048-1054.	
	6.	POPOVIC, M. et al., (2002), "Fine mapping of the locus for Shwachman-Diamond syndrome at 7q11, identification of shared disease haplotypes, and exclusion of TPST1 as a candidate gene", Eur. J. Hum. Genet, v. 10, pp. 250-8.	
	7.	KOONIN, E.V., Wolf, Y.I. & Aravind, L. (2001), "Prediction of the archaeal exosome and its connections with the proteasome and the translation and transcription machineries by a comparative-genomic approach". Genome Res. 11, 240-252.	
	8.	ROESLER, J. et al., (2000), "Recombination events between the p47-phox gene and its highly homologous pseudogenes are the main cause of autosomal recessive chronic granulomatous disease". Blood., v. 15, pp. 2150-2156.	
	9.	STRACHAN T., (1994), "Molecular pathology of 21-hydroxylase deficiency", J. Inherit. Metab. Dis., v. 17, pp. 430-41	
	10.	NEW, M.I., (1994), "Steroid 21-hydroxylase deficiency (congenital adrenal hyperplasia)", Am. J. Med., v. 98(1A), pp. 1A-2S-1A-8S.	
	11.	BEUTLER, E., (1993), "Gaucher disease as a paradigm of current issues regarding single gene mutations of humans", Proc. Natl. Acad. Sci. USA., v. 90, pp. 5384-5390.	
	12.	EIKENBOOM, J.C., et al., (1994), "Multiple substitutions in the von Willebrand factor gene that mimic the pseudogene sequence", Proc. Natl. Acad. Sci. USA., v. 91, pp. 2221-2224.	
	13.	WATNICK, T. J., et al., (1998), "Gene conversion is a likely cause of mutation in PKD1", Hum. Mol. Genet., v. 7, pp. 1239-1243.	
	14.	CHEN, J. M. and FEREC, C., (2000), "Molecular basis of hereditary pancreatitis", Eur. J. Hum. Genet., v. 8, pp. 473-479.	
	15.	CHEN, J. M., et al., (2000), "A CGC>CAT gene conversion-like event resulting in the R122H mutation in the cationic trypsinogen gene and its implication in the genotyping of pancreatitis", J. Med. Genet., v. 37, E36.	
	16.	CAI, L. et al., (2001), "A novel Q378X mutation exists in the transmembrane transporter protein ABCC6 and its pseudogene: implications for mutation analysis in pseudoxanthoma elasticum", J. Mol. Med., v. 79, pp. 536-546.	
	17.	BUNGE, S., et al., (1998), "Homologous nonallelic recombinations between the iduronate-sulfatase gene and pseudogene cause various intragenic deletions and inversions in patients with mucopolysaccharidosis type II", Eur. J. Hum. Genet., v. 6, pp. 492-500.	
	18.	HAHNEN, E., et al., (1996), "Hybrid survival motor neuron genes in patients with autosomal recessive spinal muscular atrophy: new insights into molecular mechanisms responsible for the disease", Am. J. Hum. Genet., v. 59, pp. 1057-1065.	
	19.	CAMPBELL, L., et al., (1997), "Genomic variation and gene conversion in spinal muscular atrophy: Implications for disease process and clinical phenotype", Am. J. Hum. Genet., v. 61, pp. 40-50.	
	20.	WIRTH, B., et al., (1997), "De novo rearrangements found in 2% of index patients with spinal muscular atrophy: Mutational mechanisms, parental origin, mutation rate, and implications for genetic counseling", Am. J. Hum. Genet., v. 61, pp. 1102-1111.	
	21.	ZHU, H., et al., (2001), "Global analysis of protein activities using proteome chips", Science., v. 293, pp. 2101-2105.	
	22.	BATEMAN, A., et al., (2002), "The Pfam Protein Families Database", Nucl. Acids Res. 30(1):276-280.	

Examiner Signature	/David Thomas/	Date Considered	05/27/2008
*EXAMINER: Initial if reference considered, whether or not citation is in conformance with MPEP 609. Draw line through citation if not in conformance and not considered. Include copy of this form with next communication to applicant.			

INFORMATION DISCLOSURE  
STATEMENT BY APPLICANT

(use as many sheets as necessary)

Sheet 3 of 3



Complete if Known	
Application Number	10/526,429
Filing Date	February 28, 2005
First Named Inventor	ROMMENS, Johanna M.
Group Art Unit	TBD
Examiner Name	TBD
Attorney Docket Number	8092-002-US

## OTHER NON PATENT LITERATURE DOCUMENTS

Examiner Initials*	Cite No.	Include name of the author (in CAPITAL LETTERS), title of the article (when appropriate), title of the item (book, magazine, journal, serial, symposium, catalog, etc.), date, page(s), volume-issue number(s), publisher, city and/or country where published	T
/D.T./	23.	WINZELER, E. A., et al., (1999), "Functional characterization of the <i>S. cerevisiae</i> genome by gene deletion and parallel analysis", <i>Science</i> , v. 285, pp. 901-906.	
	24.	WU, L. F. et al., (2002), "Large-scale prediction of <i>Saccharomyces cerevisiae</i> gene function using overlapping transcriptional clusters", <i>Nat. Genet.</i> , v. 31, pp. 255-265.	
	25.	MILLER, S.A., DYKES, D.D., & POLESKY, H.F. (1988), "A simple salting out procedure for extracting DNA from human nucleated cells", <i>Nucleic Acids Res.</i> v. 16, 1215.	
	26.	MACDONALD, R.J., et al., (1987) "Isolation of RNA using guanidinium salts", <i>Meth. Enzymol.</i> 152, 219-234.	
	27.	BENSON, D.A., et al., (2002), <i>GenBank</i> . <i>Nucleic Acids Res.</i> v. 30, 17-20.	
	28.	HUBBARD, T., et al., (2002), "The Ensembl genome database project", <i>Nucleic Acids Res.</i> 30, 38-41.	
	29.	SCHWARTZ, S., et al. (2000), "PipMaker – A Web Server for Aligning Two Genomic DNA Sequences", <i>Genome Res.</i> v. 10, 577-586.	
	30.	ROZEN, S. and SKALETSKY, H.J., (2000), "Primer3 on the WWW for general users and for biologist programmers", In: Krawetz, S., and S. Misener. <i>Bioinformatics Methods and Protocols: Methods in Molecular Biology</i> , Humana Press, Totowa, NJ.	
	31.	ISOGAI, T., et al. (2000), "NEDO human cDNA sequencing project", <i>Genbank</i> Accession no. AK001779, Abstract XP002268960.	
	32.	POPOVIC, M., et al., (2000), "Refined mapping of the Shwachman-Diamond syndrome locus at 7p12-q11" <i>Amer. J. Human Genetics</i> v. 67, pp. 321: XP002268959: Abstract.	
	33.	BOOCOCK, G., et al., (2003), "Mutations in SBDS are associated with Shwachman-Diamond syndrome" <i>Nature Genetics</i> , v. 33, pp. 97-101.	
	34.	PRADES, C., et al., (2002), "The human ATP binding cassette gene ABCA13, located on chromosome 7P12.3, encodes a 5058 amino acid protein with an extracellular domain encoded in part by a 4.8-kb conserved exon", <i>Cytogenetic and Genome Research</i> v. 98(2-3):160-168.	
	35.	SMITH, A., et al., (2002), "Intermittent 20q-and consistent i(7q) in a patient with Shwachman-Diamond syndrome," <i>Pediatr. Hematol. Oncol.</i> v. 19(7):525-528.	
	36.	ELGHETANY, M.T. and ALTER, B.P., (2002) "p53 Protein Overexpression in Bone Marrow biopsies of patients with Shwachman-Diamond syndrome has a prevalence similar to that of patients with refractory anemia", <i>Arch. Pathol. Lab. Med.</i> v. 126(4):452-455.	
	37.	JELIC, T.M., et al., (2001) "Expression of CD5 on Hematogones in a 7-year-old girl with Shwachman-Diamond Syndrome", <i>Pediatr. Develop. Pathol.</i> v. 4(5):505-511.	
	38.	SPIRITO, F.R., et al., (2000) "Cytogenetic characterization of acute myeloid leukemia in Shwachman's syndrome, A case report, <i>Haematologica</i> v. 85(11):1207-1210.	
	39.	DALE, D.C., et al., (2000) "Mutations in the gene encoding neutrophil elastase in congenital and cyclic neutropenia", <i>Blood</i> , v. 96(7):2317-2322.	
▼	40.	SOKOLIC, R.A., et al., (1999) "Discordant detection of monosomy 7 by GTG-banding and FISH in a patient with Shwachman-Diamond syndrome without evidence of myelodysplastic syndrome or acute myelogenous leukemia", <i>Cancer Genetics &amp; Cytogenetics.</i> , 115(2):106-13.	

Examiner Signature	/David Thomas/	Date Considered	05/27/2008
--------------------	----------------	-----------------	------------

\*EXAMINER: Initial if reference considered, whether or not citation is in conformance with MPEP 609. Draw line through citation if not in conformance and not considered. Include copy of this form with next communication to applicant.